WHAT IS CLAIMED IS:

1. An isolated DNA comprising nucleic acid of SEQ ID NO:1.

2. The isolated DNA of claim 1 wherein said DNA comprises a mutation selected from: an A at base 664, an A at base 1102, a G at base 1106, a C at base 1116, a C at base 1220, a T at base 1258, a deletion of bases 662-664, a C at base 694, an A at base 727, an A at base 731, an A at base 922, a T at base 979, an A at base 1078, a T at base 1097, an A at base 1184, a T at base 1184 or an A at base 1196.

3. An isolated DNA comprising DNA encoding a mutant KVLQT1 polypeptide which causes long QT syndrome wherein said isolated DNA comprises a mutation wherein said mutation results in said isolated DNA encoding KVLQT1 of SEQ ID NO:2 with an altered amino acid selected from the group consisting of: an Arg at position 168, a Ser at position 314, a Cys at position 315, an Asn at position 318, a Pro at position 353, a Trp at position 366, a Trp at position 167 concurrent with a deletion of amino acid residue 168, a Pro at position 178, an ARG at position 189, a Gln at position 190, a Met at position 254, a Phe at position 273, an Arg at position 306, an Ile at position 312, a Glu at position 341, a Val at position 341 or a Glu at position 345.

4. A nucleic acid probe which hybridizes specifically to the DNA of claim 2 under stringent hybridization conditions wherein said stringent hybridization conditions prevent said nucleic acid probe from hybridizing to DNA of SEQ ID NO:1.

5. A nucleic acid probe which hybridizes specifically to the DNA of claim 3 under stringent hybridization conditions wherein said stringent hybridization conditions prevent said nucleic acid probe from hybridizing to DNA of SEQ ID NO:1.

6. A method for diagnosing a polymorphism which causes long QT syndrome comprising hybridizing a probe of claim 4 to a patient's sample of DNA or RNA under stringent conditions which allow hybridization of said probe to nucleic acid comprising said polymorphism but

prevent hybridization of said probe to wild-type *KVLQT1* wherein the presence of a hybridization signal indicates the presence of said polymorphism.

7. A method for diagnosing a polymorphism which causes long QT syndrome comprising hybridizing a probe of claim 5 to a patient's sample of DNA or RNA under stringent conditions which allow hybridization of said probe to nucleic acid comprising said polymorphism but prevent hybridization of said probe to wild-type *KVLQT1* wherein the presence of a hybridization signal indicates the presence of said polymorphism.

8. The method according to claim 6 wherein the patient's DNA or RNA has been amplified and said amplified DNA or RNA is hybridized.

9. The method according to claim 7 wherein the patient's DNA or RNA has been amplified and said amplified DNA or RNA is hybridized.

10. A method according to claim 8 wherein hybridization is performed *in situ*.

11. A method according to claim 9 wherein hybridization is performed *in situ*.

12. A method for diagnosing the presence of a polymorphism in human *KVLQT1* which causes long QT syndrome wherein said method is performed by means which identify the presence of said polymorphism, wherein said polymorphism is one which results in the presence of a KVLQT1 polypeptide of SEQ ID NO:2 with an altered amino acid, selected from the group consisting of: an Arg at residue 168, a Ser at residue 314, a Cys at residue 315, an Asn at residue 318, a Pro at residue 353, a Trp at residue 366, a Trp at position 167 concurrent with a deletion of amino acid residue 168, a Pro at position 178, an ARG at position 189, a Gln at position 190, a Met at position 254, a Phe at position 273, an Arg at position 306, an Ile at position 312, a Glu at position 341, a Val at position 341 or a Glu at position 345.

13. The method of claim 12 wherein said polymorphism is the presence of an A at base 664, an A at base 1102, a G at base 1106, a C at base 1116, a C at base 1220, a T at base 1258, a deletion of bases 662-664, a C at base 694, an A at base 727, an A at base 731, an A at base 922, a T at base 979, an A at base 1078, a T at base 1097, an A at base 1184, a T at base 1184 or an A at base 1196.

14. The method of claim 12 wherein said means comprises using a single-stranded conformation polymorphism technique to assay for said polymorphism.

15. The method of claim 12 wherein said means comprises sequencing human *KVLQT1*.

16. The method of claim 12 wherein said means comprises performing an RNAse assay.

17. The method of claim 13 wherein said means comprises using a single-stranded conformation polymorphism technique to assay for said polymorphism.

18. The method of claim 13 wherein said means comprises sequencing human *KVLQT1*.

19. The method of claim 13 wherein said means comprises performing an RNAse assay.

20. An antibody which binds to a mutant KVLQT1 polypeptide but not to wild-type KVLQT1 polypeptide, wherein said mutant KVLQT1 polypeptide comprises an Arg at residue 168, a Ser at residue 314, a Cys at residue 315, an Asn at residue 318, a Pro at residue 353, a Trp at residue 366, a Trp at position 167 concurrent with a deletion of amino acid residue 168, a Pro at position 178, an ARG at position 189, a Gln at position 190, a Met at position 254, a Phe at position 273, an Arg at position 306, an Ile at position 312, a Glu at position 341, a Val at position 341 or a Glu at position 345.

21. A method for diagnosing long QT syndrome said method consisting of an assay for the presence of mutant KVLQT1 polypeptide in a patient by reacting a patient's sample with an antibody of claim 20 wherein the presence of a positive reaction is indicative of long QT syndrome.

22. The method of claim 21 wherein said antibody is a monoclonal antibody.

23. The method of claim 21 wherein said assay comprises immunoblotting.

24. The method of claim 21 wherein said assay comprises an immunocytochemical technique.

25. An isolated KVLQT1 polypeptide comprising a mutation which causes long QT syndrome wherein said mutation is an Arg at residue 168, a Ser at residue 314, a Cys at residue 315, an Asn at residue 318, a Pro at residue 353, a Trp at residue 366, a Trp at position 167 concurrent with a deletion of amino acid residue 168, a Pro at position 178, an ARG at position 189, a Gln at position 190, a Met at position 254, a Phe at position 273, an Arg at position 306, an Ile at position 312, a Glu at position 341, a Val at position 341 or a Glu at position 345.

26. A method for diagnosing long QT syndrome in a person wherein said method comprises sequencing a KVLQT1 polypeptide from said person or sequencing KVLQT1 polypeptide synthesized from nucleic acid derived from said person wherein the presence of an Arg at residue 168, a Ser at residue 314, a Cys at residue 315, an Asn at residue 318, a Pro at residue 353, a Trp at residue 366, a Trp at position 167 concurrent with a deletion of amino acid residue 168, a Pro at position 178, an ARG at position 189, a Gln at position 190, a Met at position 254, a Phe at position 273, an Arg at position 306, an Ile at position 312, a Glu at position 341, a Val at position 341 or a Glu at position 345 is indicative of long QT syndrome.

27. An isolated nucleic acid selected from the group consisting of SEQ ID NO:41, SEQ ID NO:42, SEQ ID NO:43, SEQ ID NO:44, SEQ ID NO:45, SEQ ID NO:46, SEQ ID NO:47, SEQ ID NO:48, SEQ ID NO:49, SEQ ID NO:50, SEQ ID NO:51, SEQ ID NO:52, SEQ ID NO:53, SEQ ID NO:54, SEQ ID NO:55, SEQ ID NO:56, SEQ ID NO:57, SEQ ID NO:58,

SEQ ID NO:59, SEQ ID NO:60, SEQ ID NO:61, SEQ ID NO:62, SEQ ID NO:63, SEQ ID NO:64, SEQ ID NO:65, SEQ ID NO:66, SEQ ID NO:67, SEQ ID NO:68, SEQ ID NO:69, SEQ ID NO:70, SEQ ID NO:71, SEQ ID NO:72, SEQ ID NO:73 and SEQ ID NO:74.

28. A pair of nucleic acid primers wherein said primers are:

a) SEQ ID NOs:41 and 42;
b) SEQ ID NOs:43 and 44;
c) SEQ ID NOs:45 and 46;
d) SEQ ID NOs:47 and 48;
e) SEQ ID NOs:49 and 50;
f) SEQ ID NOs:51 and 52;
g) SEQ ID NOs:53 and 54;
h) SEQ ID NOs:55 and 56;
i) SEQ ID NOs:57 and 58;
j) SEQ ID NOs:59 and 60;
k) SEQ ID NOs:61 and 62;
l) SEQ ID NOs:63 and 64;
m) SEQ ID NOs:65 and 66;
n) SEQ ID NOs:67 and 68;
o) SEQ ID NOs:69 and 70;
p) SEQ ID NOs:71 and 72; or
q) SEQ ID NOs:73 and 74.

29. A method of amplifying an exon of *KVLQT1* wherein said method comprises using a pair of primers.

30. A method of amplifying an exon of *KVLQT1* wherein said method comprises using a pair of primers selected from the primer pairs of claim 28.

31. A method to screen for drugs which are useful in treating a person with a mutation in *KVLQT1,* wherein said mutation is one which results in an Arg at amino acid residue 168, a Ser at amino acid residue 314, a Cys at amino acid residue 315, an Asn at amino acid residue 318, a Pro at amino acid residue 353, a Trp at amino acid residue 366, a Trp at position 167 concurrent with a deletion of amino acid residue 168, a Pro at position 178, an ARG at position 189, a Gln at position 190, a Met at position 254, a Phe at position 273, an Arg at position 306, an Ile at position 312, a Glu at position 341, a Val at position 341 or a Glu at position 345 said method comprising:

a) placing a first set of cells expressing KVLQT1 with a mutation, wherein said mutation is an Arg at amino acid residue 168, a Ser at amino acid residue 314, a Cys at amino acid residue 315, an Asn at amino acid residue 318, a Pro at amino acid residue 353, a Trp at amino acid residue 366, a Trp at position 167 concurrent with a deletion of amino acid residue 168, a Pro at position 178, an ARG at position 189, a Gln at position 190, a Met at position 254, a Phe at position 273, an Arg at position 306, an Ile at position 312, a Glu at position 341, a Val at position 341 or a Glu at position 345 into a bathing solution to measure a first induced K^+ current;

b) measuring said first induced K^+ current;

c) placing a second set of cells expressing wild-type KVLQT1 into a bathing solution to measure a second induced K^+ current;

d) measuring said second induced K^+ current;

e) adding a drug to the bathing solution of step (a);

f) measuring a third induced K^+ current of cells in step (e); and

g) determining whether the third induced K^+ current is more similar to the second induced K^+ current than is the first induced K^+ current, wherein drugs resulting in a third induced K^+ current which is closer to the second induced K^+ current than is the first induced K^+ current are useful in treating said persons.

32. The method of claim 31 wherein said first set of cells or said second set of cells is obtained from a transgenic animal.

33. An isolated nucleic acid or its complement encoding a polypeptide of SEQ ID NO:2.

34. An isolated nucleic acid comprising any 15 consecutive nucleotides of SEQ ID NO:1 or its complement.

35. An isolated nucleic acid comprising any 12 consecutive nucleotides of SEQ ID NO:1 or its complement.

Sub a3

36. An isolated nucleic acid comprising any 15 consecutive nucleotides of SEQ ID NO:1 or its complement wherein SEQ ID NO:1 comprises one or more mutations selected from the group consisting of: an A at base 664, an A at base 1102, a G at base 1106, a C at base 1116, a C at base 1220, a T at base 1258, a deletion of bases 662-664, a C at base 694, an A at base 727, an A at base 731, an A at base 922, a T at base 979, an A at base 1078, a T at base 1097, an A at base 1184, a T at base 1184 or an A at base 1196.

37. An isolated nucleic acid comprising any 12 consecutive nucleotides of SEQ ID NO:1 or its complement wherein SEQ ID NO:1 comprises one or more mutations selected from the group consisting of: an A at base 664, an A at base 1102, a G at base 1106, a C at base 1116, a C at base 1220, a T at base 1258, a deletion of bases 662-664, a C at base 694, an A at base 727, an A at base 731, an A at base 922, a T at base 979, an A at base 1078, a T at base 1097, an A at base 1184, a T at base 1184 or an A at base 1196.

38. A cell transfected with the DNA of claim 1.

39. A cell transfected with the DNA of claim 2.

40. A cell transfected with the DNA of claim 3.

41. A cell transfected with the nucleic acid of claim 33.

42. A vector comprising the isolated DNA of claim 1.

43. A vector comprising the isolated DNA of claim 2.

44. A vector comprising the isolated DNA of claim 3.

45. A vector comprising the isolated nucleic acid of claim 33.

46. A cell transfected with the vector of claim 42.

47. A cell transfected with the vector of claim 43.

48. A cell transfected with the vector of claim 44.

49. A cell transfected with the vector of claim 45.

50. A nonhuman, transgenic animal comprising the DNA of claim 1.

51. A nonhuman, transgenic animal comprising the DNA of claim 2.

52. A nonhuman, transgenic animal comprising the DNA of claim 3.

53. A nonhuman, transgenic animal comprising the nucleic acid of claim 33.

54. An isolated polypeptide of SEQ ID NO:2.

55. An antibody which specifically binds to the polypeptide of claim 46.

56. A method of assessing a risk in a human subject for long QT syndrome which comprises screening said subject for a mutation in a KVLQT1 gene by comparing the sequence of the KVLQT1 gene or its expression products isolated from a tissue sample of said subject with a wild-type KVLQT1 gene or its expression products, wherein a mutation in the sequence of the subject is indicative of a risk for long QT syndrome.

57. The method of claim 56 wherein said expression product is selected form the group consisting of mRNA of the KVLQT1 gene and a KVLQT1 polypeptide encoded by the KVLQT1 gene.

58. The method of claim 56 wherein one or more of the following procedures is carried out:

(a) observing shifts in electrophoretic mobility of single-stranded DNA from said sample on non-denaturing polyacrylamide gels;

(b) hybridizing a KVLQT1 gene probe to genomic DNA isolated from said sample under conditions suitable for hybridization of said probe to said gene;

(c) determining hybridization of an allele-specific probe to genomic DNA from said sample;

(d) amplifying all or part of the KVLQT1 gene from said sample to produce an amplified sequence and sequencing the amplified sequence;

(e) determining by nucleic acid amplification the presence of a specific KVLQT1 mutant allele in said sample;

(f) molecularly cloning all or part of the KVLQT1 gene from said sample to produce a cloned sequence and sequencing the cloned sequence;

(g) determining whether there is a mismatch between molecules (1) KVLQT1 gene genomic DNA or KVLQT1 mRNA isolated from said sample, and (2) a nucleic acid probe complementary to the human wild-type KVLQT1 gene DNA, when molecules (1) and (2) are hybridized to each other to form a duplex;

(h) amplification of KVLQT1 gene sequences in said sample and hybridization of the amplified sequences to nucleic acid probes which comprise wild-type KVLQT1 gene sequences;

(i) amplification of KVLQT1 gene sequences in said tissue and hybridization of the amplified sequences to nucleic acid probes which comprise mutant KVLQT1 gene sequences;

(j) screening for a deletion mutation;

(k) screening for a point mutation;

(l) screening for an insertion mutation;

(m) determining *in situ* hybridization of the KVLQT1 gene in said sample with one or more nucleic acid probes which comprise the KCNE1 gene sequence or a mutant KVLQT1 gene sequence;

(n) immunoblotting;

(o) immunocytochemistry;

(p) assaying for binding interactions between KVLQT1 gene protein isolated from said tissue and a binding partner capable of specifically binding the polypeptide expression product of a KVLQT1 mutant allele and/or a binding partner for the KVLQT1 polypeptide having the amino acid sequence set forth in SEQ ID NO:2; and

(q) assaying for the inhibition of biochemical activity of said binding partner.

59. An isolated nucleic acid encoding a *Xenopus* KVLQT1 polypeptide having the amino acid sequence set forth in SEQ ID NO:113.

60. An isolated *Xenopus* polypeptide having the amino acid sequence set forth in SEQ ID NO:113.

61. An isolated nucleic acid comprising any 15 consecutive nucleotides of the nucleic acid of claim 59 or its complement.

62. An isolated nucleic acid comprising any 12 consecutive nucleotides of the nucleic acid of claim 59 or its complement.

63. An antibody which specifically binds to the polypeptide of claim 60.